Speaker Profiles – National OI Conference Sydney October 2019

Professor Sillence

AM, MD(Melb), MBBS(Syd), FRACP, FRCPA, FAFPHM, FAFRM ,FFSci (RCPA) | Clinical Geneticist

Professor Sillence is the foundation chair of Medical Genetics in the University of Sydney. An honours graduate of the University of Sydney, he obtained his MD in Medical Genetics from the University of Melbourne 1978.

He serves on the education committee of the Human Genetics Society of Australasia, the International Nomenclature Committee for Constitutional Disorders of the Skeletal, the International Mucopolysaccharidosis type I expert committee, the National Fabry Disease and MPS expert committees for the LSDP. David Sillence's current research interests include:

a) Genetics and treatment of osteopenic and other metabolic bone disorders of childhood,

b) Characterization of the molecular genetics and pathogenesis of specific skeletal birth defects in mouse and man,

c) Consanguinity and paediatric morbidity/population genetics of consanguinity in Middle Eastern Populations,

d) Evaluation of Innovative Genetic Therapies.

These studies are being undertaken with a range of collaborators from within the Children's Hospital at Westmead, the research institutes in Sydney and with overseas geneticists. Our studies in the genetics and treatment of osteopenic and other metabolic bone disorders has led to the development of:

i) Normal range of bone density and skeletal metabolise in children,

ii) a delineation of the natural history of various skeletal disorders collectively known as Osteogenesis Imperfecta and

iii) the definition of the specific conditions for treatment of these disorders with Bisphosphonates.

Professor Sillence has also worked closely with the Victor Chang Developmental Biology Unit at the Garvan Institute in Sydney in developing an approach to studying congenital anomalies of spine development.

Professor Sillence has over considerable period developed studies related to consanguinity and paediatric morbidity. Future research involves a detailed analysis of the disorders in specific populations. This will allow us to develop population screening technologies and so be able to offer couples accurate population specific genetic testing in the future. Our approaches include Autozygosity Mapping to characterize rare autosomal recessive disorders in the client populations. The collaborative group is developing a confidential register of information about rare disorders in these populations.

David Sillence also formed the centre for the evaluation of Innovative Genetic Therapies at the Westmead Hospital and the Children's Hospital at Westmead to evaluate innovative therapies such as Enzyme Replacement and Substrate Reduction Therapies in the treatment of Lysosomal Storage Disorders in Adults and Children.

Paediatric Endocrinology – National protocol and what the future holds

Professor Craig Munns

Prof Munns is a Senior Staff Specialist in Endocrinology, and Clinical Program Director for the Division of Diagnostic Services at the Children's Hospital at Westmead. He is the Professor of Paediatric Bone and Mineral Medicine at the Sydney Medical School at the University of Sydney and is Head of Clinical Research at the Centre for Children's Bone and Musculoskeletal Health, at Kids Research.

Prof Munns' primary clinical and research interest is the diagnosis and management of primary and secondary paediatric bone disorders

Physiotherapy & Connective Tissue research

Assoc. Professor Verity Pacey

Verity is an Associate Professor and Head of the Department of Health Professions within the Faculty of Medicine and Health Sciences, Macquarie University. She has almost 20 years of experience as a clinical physiotherapist, previously working as a senior physiotherapist, and then Bone and Mineral Coordinator, at The Children's Hospital at Westmead Connective Tissue Dysplasia Clinic. She supervises both students and clinicians undertaking higher degree research in various aspects of physiotherapy practice.

Verity is considered an international leader in the physiotherapy management of individuals with Connective Tissue Dysplasias, including Osteogenesis Imperfecta, and is an active member of the Short Statured People's Association of Australia Health Professional Advisory Board and Care 4 Brittle Bones Rehabilitation Group.

Subsequently, Verity is an author on international rehabilitation consensus guidelines for Osteogenesis Imperfecta published in 2018. She is an invited speaker at both national and international level within her area of expertise: rare and complex conditions in paediatrics, focusing on physical activity, function and quality of life.

Transition and Adult Clinics for OI

Professor Jenny Gunton

Professor Jenny Gunton heads the Centre for Diabetes, Obesity and Endocrinology Research (CDOER) and Westmead Institute for Medical Research (WIMR). She is Chair of Medicine at Westmead Hospital and a clinical endocrinologist / diabetologist.

She received her PhD from the University of Sydney in 2003 having studied in Rob Baxter's lab at the Kolling Institute.

She completed her post-doctoral fellowship in Ron Kahn's lab at the Joslin Diabetes Center and Harvard Medical School in 2005. She returned to Australia to the Garvan Institute and set up the Diabetes and Transcription Factors lab. In 2012 she became the President of the Australian Diabetes Society and in 2014 became Chair of Medicine at Sydney University, Westmead Hospital. Her lab moved to the new WIMR in late 2014.

	Her research interests include diabetes, obesity, and vitamin D. She is particularly interested in the intersection of transcription factors and their regulation by nutrients.
International Research Adult OI - Endocrinology 	***Dr Jannie Dahl Hald MD, PhD*** *Jannie Dahl Hald is a Danish adult endocrinologist working at Department of Endocrinology and Internal Medicine at Aarhus University Hospital.* *During 2019 located in Sydney where she is doing an observership at Childrens Hospital Westmead. She has a particular interest in genetic bone disorders and has done her PhD within adult osteogenesis imperfecta. She conducted a large cross-sectional study investigating bone phenotype as well as hearing loss, dental implications, eye issues and quality of life in an adult OI population.* *The need for follow up and care in adulthood will be discussed based on the results of these studies – with a reflection on differences between countries. The above studies are conducted in close collaboration with the Danish OI society.*
Respiratory Care for Adults with OI	***Dr Brist Roy - Respiratory and Sleep Physician***
Dental Care and Risk of Medications in OI Children and Young Adults	***Dr Sally Hibbert*** *Sally is employed as a Staff Specialist in Paediatric Dentistry at the Children's Hospital at Westmead and Westmead Centre for Oral Health. She was born in the UK and completed her BDS at Liverpool in 1986.* *Having completed her Fellowship at the Royal College of Surgeons Edinburgh in 1993, she was employed as lecturer in Paediatric Dentistry at the University of Liverpool, a post that she held until moving to Sydney in 2002. During that time she completed a Masters degree, together with the UK Specialist Training Pathway in Paediatric Dentistry and was awarded the Intercollegiate Speciality Fellowship in 2002.*

	*Sally has lectured widely in the UK, Ireland and Australasia on the issues of management of anxiety in children and use of hypnosis, dental trauma and care of the primary dentition.* *Current research interests include the epidemiology and planning of out-of-hours dental emergencies and the dental management of children with complex medical co-morbidities.*
Occupational Therapy & Connective Tissue research	***Alison Wesley*** *Alison Wesley is a Senior Occupational Therapist at The Children's Hospital at Westmead, Sydney, Australia. She has had over 20 years working as a paediatric Occupational Therapist. In the last 12 years she has specialised in working with children with Heritable Disorder of Connective Tissue and Skeletal Dysplasia including children with Osteogenesis Imperfecta. She has a special interest in the management of hand function for children with these conditions.* *Alison holds a Bachelor of Science through University of NSW, a Graduate Diploma of Occupational Therapy through University of Sydney, and a Master of Education through University of NSW.* *Alison is pursuing her interest in the upper limb management of children with connective tissue dysplasia through a PhD. Her research involves looking at the effect on hand use of characteristics of a connective tissue dysplasia on the daily function of children.*
Spine Care	***Dr Randolph Gray***
NDIS – Barriers to Entry	***Bernadette Saberton & Julie Haraksin***